Chromosome Structure Changes

by Sophia

WHAT'S COVERED

In this lesson, you will learn to identify types of chromosome structure changes. Specifically, this lesson will cover:

1. Gene Mutations

A gene mutation is the change in the nucleotides that make up a gene. Sometimes, chromosome structure can be altered during cell division, which will lead to a certain type of gene mutation. These gene mutations can include:

- Deletions
- Translocations
- Duplications

1a. Deletions

A **deletion** is the removal of one or more nucleotides on the chromosome.

↱ EXAMPLE **Cri-du-chat** is a disorder that is caused by a deletion on chromosome number five. With this type of disorder, a baby's larynx will not properly develop, so that when the baby cries, it will sound like a meowing cat. This disorder can also be associated with abnormal mental development as well.

TERMS TO KNOW

Deletion

When part of a chromosome is deleted; sometimes it may not have any effect, while other times it does.

Cri-du-chat

A genetic disorder caused by the deletion of a section of the fifth chromosome; cri-du-chat is French for “cry of the cat” and causes physical and cognitive abnormalities.

1b. Translocations

Translocation is when part of one chromosome switches place with a corresponding part of a different (non-homologous) chromosome. This type of gene mutation can lead to certain types of cancers.

↱ EXAMPLE Say the original Chromosome 1 is made of A, B, C, D, E, F and Chromosome 2 contains G, H, I, J, K, L. In a translocation, part of Chromosome 1 will switch places with a corresponding part of Chromosome 2. In this example, perhaps the F and L parts of the chromosome could switch places so that the translocated Chromosome 1 now reads ABCDEL and the translocated Chromosome 2 now reads GHIJKF.

TERM TO KNOW

Translocation

A chromosome abnormality that occurs when pieces of different (non-homologous) chromosomes fuse together.

1c. Duplication

Duplication is when a nucleotide sequence is repeated. Duplications can be harmful, as in the case of Huntington's disease. The more repeats of a certain nucleotide sequence you have, the more likely you are to contract Huntington's; an increase in this particular duplication also means you'll contract Huntington's earlier in life. However, duplications can also be helpful. Our ancestors used to be unable to see green and yellow color. A duplication of our red cone cell receptor gene, along with some slight mutation, resulted in a third type of cone cell in our retina. This duplication allows us to see green and yellow light, as well as blue and red.

↱ EXAMPLE Say a chromosome is composed of A, B, C, D, E, F. If a duplication occurs, it might end up with A, B, C, B, C, D, E, F with the B, C portion repeated.

TERM TO KNOW

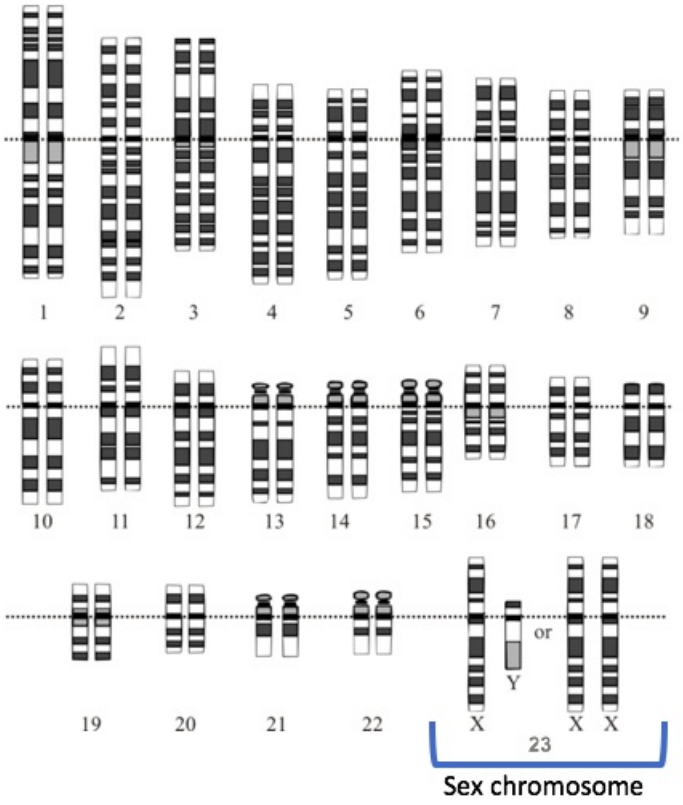
Duplication

Sequences of nucleotides are repeated multiple times. Huntington's disease is an example of a genetic disorder that results from duplication: the more times a certain sequence is repeated, the more likely and earlier in life Huntington's is to strike.

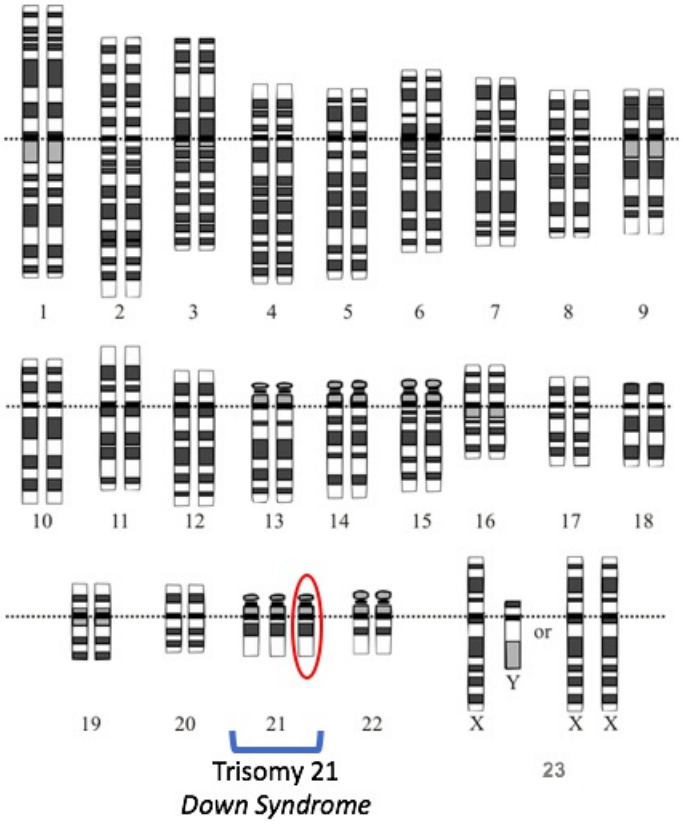
---

2. Karyotypes

Karyotypes are the arrangement of a person's complete set of chromosomes by length, shape, and the location of the centromere. These pictures are taken well in metaphase of mitosis because the chromosomes at that point are most easy to identify. The chromosomes will be photographed through the microscope, cut out, and arranged from the largest all the way down to the smallest. The last pair of chromosomes in a karyotype are always the sex chromosomes. For a male, one is an X chromosome, and one is the Y chromosome. For a female, they will both be X chromosomes.



These karyotypes allow you to see a picture of a person's chromosome. This can help to identify any abnormalities. One abnormality that can be seen on a karyotype is Trisomy 21, a genetic disorder in which a person has three copies of chromosome number 21. Here is an example of what this might look like:



If there is a third chromosome in the 21st pair, then the person would have trisomy 21; also known as Down syndrome.

TERM TO KNOW

Karyotype
Looking at the number of chromosomes and their characteristics under a microscope; this is a critical tool in assessing genetic disorders in a developing embryo.

SUMMARY

Gene mutations are changes in the nucleotide sequence. These mutations can include**deletions**, **translocations**, and **duplications**. A deletion is the removal of one or more nucleotides on the chromosome. Translocation is when part of a chromosome switches places with part of a different (non-homologous) chromosome. Duplication occurs when part of a chromosome becomes repeated. A **karyotype** looks at a person's chromosomes under a microscope to check for genetic abnormalities. This can identify if someone has Down syndrome.

Keep up the learning and have a great day!

Source: THIS WORK IS ADAPTED FROM SOPHIA AUTHOR AMANDA SODERLIND

ATTRIBUTIONS

- Karotype | **Author:** Wikipeda | **License:** Public Domain
- Trisomy 21 | **Author:** Wikipeda | **License:** Public Domain

TERMS TO KNOW

Cri-du-chat
A genetic disorder caused by the deletion of a section of the fifth chromosome; cri-du-chat is French for “cry of the cat”. Cri-du-chat causes physical and cognitive abnormalities.

Deletion
When part of a chromosome is deleted; sometimes it may not have any effect while other times it does.

Duplication
Sequences of nucleotides are repeated multiple times. Huntington's disease is an example of a genetic disorder that results from duplication: the more times a certain sequence is repeated, the more likely and earlier in life Huntington's is to strike.

Karyotype
Looking at the number of chromosomes and their characteristics under a microscope. This is a critical tool in assessing genetic disorders in a developing embryo.

Translocation
A chromosome abnormality that occurs when pieces of different (non-homologous) chromosomes fuse together.